

ABSTRACT

The present invention provides an efficient way for high throughput haplotype analysis. Several polymorphic nucleic acid markers, such as SNPs, can be simultaneously and reliably determined through multiplex PCR of single nucleic acid molecules in several parallel single molecule dilutions and the consequent statistical analysis of the results from these parallel single molecule multiplex PCR reactions results in reliable determination of haplotypes present in the subject. The nucleic acid markers can be of any distance to each other on the chromosome. In addition, an approach wherein overlapping DNA markers are analyzed can be used to link smaller haplotypes into larger haplotypes. Consequently, the invention provides a powerful new tool for diagnostic haplotyping and identifying novel haplotypes.